

Table S2. 86 germline nonsynonymous mutations were identified in blood sample of patient-1 by Exome sequencing.

#Symbol	Location	Transcript	Protein_position	Amino_acids	Codons
ACOX2	3:58512313	ENST00000459701	395	R/H	cGc/cAc
ADAMTS14	10:72513635	ENST00000373208	940	L/M	Ctg/Atg
ADAP2	17:29276342	ENST00000581548	110	R/H	cGt/cAt
ADGRG4	X:135430483	ENST00000370652	1540	S/P	Tct/Cct
AHNAK	11:62303548	ENST00000528508	8	R/Q	cGg/cAg
ALCAM	3:105264151	ENST00000306107	359	S/N	aGc/aAc
ALDH4A1	1:19209799	ENST00000538309	133	N/H	Aac/Cac
ANKRD36	2:97877478	ENST00000461153	1157	V/M	Gtg/Atg
ANXA9	1:150958836	ENST00000368947	166	D/G	gAc/gGc
ARPIN	15:90446572	ENST00000560096	70	C/Y	tGt/tAt
ATP6V1E2	2:46739684	ENST00000306448	56	Y/C	tAt/tGt
BRCA2	13:32911755	ENST00000544455	1088	P/H	cCt/cAt
BTN3A3	6:26451927	ENST00000490254	138	T/M	aCg/aTg
CA2	8:86392912	ENST00000285379	226	R/H	cGt/cAt
CEP78	9:80879095	ENST00000376598	496	N/K	aaT/aaA
CFAP54	12:96900715	ENST00000298953	193	L/V	Ttg/Gtg
CTSH	15:79227412	ENST00000528741	29	T/P	Acc/Ccc
CX3CR1	3:39307666	ENST00000542107	112	G/A	gGc/gCc
DDX53	X:23019317	ENST00000327968	381	M/I	atG/atT
DHODH	16:72048535	ENST00000572887	133	V/A	gTc/gCc
DNAH3	16:20966299	ENST00000261383	3636	P/H	cCc/cAc
FAN1	15:31218141	ENST00000362065	829	Q/H	caG/caC
FH	1:241665767	ENST00000366560	404	E/D	gaG/gaC
GPR176	15:40099241	ENST00000543580	86	T/A	Acc/Gcc
GPRIN2	10:46999019	ENST00000374317	47	V/M	Gtg/Atg
GPRIN2	10:46999484	ENST00000374317	202	G/W	Ggg/Tgg
GPRIN2	10:46999922	ENST00000374314	348	V/L	Gtg/Ttg
GRIK1	21:31015249	ENST00000399913	332	A/V	gCc/gTc
HELZ2	20:62198745	ENST00000467148	656	V/M	Gtg/Atg
HYDIN	16:71101200	ENST00000288168	707	T/A	Aca/Gca
HYDIN	16:71098649	ENST00000393552	342	N/D	Aat/Gat
IMMT	2:86400824	ENST00000254636	37	P/S	Cct/Tct
KIAA1683	19:18368720	ENST00000600359	892	R/H	cGt/cAt
KIAA1755	20:36850938	ENST00000435901	76	G/D	gGc/gAc
KLHL10	17:39998487	ENST00000293303	203	V/I	Gta/Ata
KRT40	17:39140272	ENST00000377755	85	F/S	tTc/tCc
LAMA4	6:112430669	ENST00000230538	1815	V/I	Gta/Ata
LAMA4	6:112457383	ENST00000522006	1112	P/R	cCt/cGt
LILRB4	19:55179364	ENST00000391736	414	Q/R	cAg/cGg
LIPH	3:185245370	ENST00000296252	177	L/R	cTc/cGc

LRRC49	15:71329621	ENST00000560369	608	R/C	Cgt/Tgt
MAP3K9	14:71197492	ENST00000553414	707	R/C	Cgc/Tgc
MBD5	2:149226897	ENST00000404807	462	S/L	tCa/tTa
METTL21B	12:58166617	ENST00000333012	37	G/E	gGg/gAg
MST1R	3:49934990	ENST00000344206	670	V/G	gTa/gGa
MXRA5	X:3238733	ENST00000217939	1665	P/S	Cca/Tca
NEB	2:152390806	ENST00000172853	5413	R/W	Cgg/Tgg
NEDD9	6:11190208	ENST00000379446	632	V/I	Gtc/Atc
NLRX1	11:119052976	ENST00000409265	843	A/V	gCg/gTg
NOX5	15:69329422	ENST00000260364	397	H/Y	Cat/Tat
NT5C3B	17:39983808	ENST00000415460	183	S/C	tCt/tGt
OR4M2	15:22368862	ENST00000332663	96	G/E	gGa/gAa
OSCAR	19:54598604	ENST00000358375	225	Y/S	tAc/tCc
PARD3	10:34637007	ENST00000346874	702	P/S	Ccc/Tcc
PDE4DIP	1:144866643	ENST00000530130	24	R/C	Cgc/Tgc
PDE4DIP	1:144994658	ENST00000369351	25	R/L	cGc/cTc
PHKB	16:47536987	ENST00000455779	124	R/C	Cgt/Tgt
PHYH	10:13330389	ENST00000263038	217	L/V	Ctg/Gtg
PSMD10	X:107328208	ENST00000217958	226	G/D	gGt/gAt
PTPRT	20:40944422	ENST00000373187	694	P/A	Cct/Gct
PZP	12:9312951	ENST00000261336	1003	T/M	aCg/aTg
RGPD3	2:107073501	ENST00000409886	111	D/N	Gat/Aat
SEC31B	10:102257953	ENST00000370345	566	L/F	Ctc/Ttc
SLC25A47	14:100792549	ENST00000361529	43	T/M	aCg/aTg
SPATA6	1:48764419	ENST00000371847	478	C/Y	tGt/tAt
SPTBN2	11:66483347	ENST00000529997	88	G/E	gGa/gAa
SRRT	7:100482950	ENST00000388793	425	R/C	Cgc/Tgc
SSX5	X:48054740	ENST00000347757	19	E/Q	Gag/Cag
TAS1R2	1:19166157	ENST00000375371	819	P/L	cCg/cTg
TEAD2	19:49852034	ENST00000539846	93	R/W	Cgg/Tgg
TMEM70	8:74888679	ENST00000517439	55	G/W	Ggg/Tgg
TMPRSS5	11:113565252	ENST00000299882	245	G/S	Ggc/Agc
TTC28	22:28394576	ENST00000397906	1691	K/E	Aag/Gag
TTC38	22:46674494	ENST00000422713	135	G/A	gGc/gCc
TTLL12	22:43568512	ENST00000216129	464	V/M	Gtg/Atg
TUBGCP6	22:50657782	ENST00000248846	1473	Q/H	caG/caT
UTP20	12:101736761	ENST00000261637	1447	D/N	Gac/Aac
VDR	12:48272895	ENST00000395324	1	M/T	aTg/aCg
VPS39	15:42454577	ENST00000348544	771	A/T	Gct/Act
WDR19	4:39241899	ENST00000399820	789	G/D	gGt/gAt
WDR34	9:131397002	ENST00000372715	394	P/S	Ccc/Tcc
WDR47	1:109547332	ENST00000357672	351	T/I	aCa/aTa
ZNF100	19:21909948	ENST00000358296	389	T/I	aCt/aTt
ZNF66	19:20989989	ENST00000344519	528	K/M	aAg/aTg

ZNF776	19:58265373	ENST00000317178	292	Y/C	tAt/tGt
ZNF90	19:20229486	ENST00000418063	375	G/S	Ggc/Agc